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Myopathology in times of modern imaging.

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Abstract

Over the last two decades muscle (magnetic resonance) imaging has become an important complementary tool in the diagnosis and differential diagnosis of inherited neuromuscular disorders, particularly in conditions where the pattern of selective muscle involvement is often more predictive of the underlying genetic background than associated clinical and histopathological features. Following an overview of different imaging modalities, the present review will give a concise introduction to systematic image analysis and interpretation in genetic neuromuscular disorders. The pattern of selective muscle involvement will be presented in detail in conditions such as the congenital or myofibrillar myopathies where muscle imaging is particularly useful to inform the (differential) diagnosis, and in disorders such as Duchenne or fascioscapulohumeral muscular dystrophy where the diagnosis is usually made on clinical grounds but where detailed knowledge of disease progression on the muscle imaging level may inform better understanding of the natural history. Utilizing the group of the congenital myopathies as an example, selected case studies will illustrate how muscle MRI can be used to inform the diagnostic process in the clinico-pathological context. Future developments, in particular concerning the increasing use of whole body MRI protocols and novel quantitative fat assessments techniques potentially relevant as an outcome measure, will be briefly outlined. This article is protected by copyright. All rights reserved.

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KEYWORDS: Congenital muscular dystrophies; Congenital myopathies; Limb girdle muscular dystrophies; Muscle magnetic resonance imaging; Myofibrillar myopathies

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